
**Event Agenda**

8:00 a.m. EST **Registration and Exhibit Booth/Poster Setup**

9:00 a.m. **Rare Disease Day at NIH Overview**
**Meera A. Shah, M.P.H.**, Health Specialist, Division of Rare Diseases Research Innovation (DRDRI), National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH)

9:10 a.m. **NIH Director Remarks**
**Jay Bhattacharya, M.D., Ph.D.**, Director, NIH

9:20 a.m. **Welcoming Remarks**
**Joni L. Rutter, Ph.D.**, Director, NCATS, NIH

9:30 a.m. **Rare Disease Congressional Caucus Remarks**
**Rep. Gus M. Bilirakis (R-FL)**, House Co-chair [video]
**Rep. Doris O. Matsui (D-CA)**, House Co-chair [video]
**Sen. Amy J. Klobuchar (D-MN)**, Senate Co-chair [video]
**Sen. Roger F. Wicker (R-MS)**, Senate Co-chair [video]

9:50 a.m. **NCATS DRDRI Update**
**Philip John (P.J.) Brooks, Ph.D.**, Acting Director, DRDRI, NCATS, NIH

10:05 a.m. **Beyond the Diagnosis Portrait Unveiling**
**Patricia Weltin**, CEO/Founder, Beyond the Diagnosis
**Lucas Kolasa**, Artist, Beyond the Diagnosis
**Aisha Campbell, M.B.A./H.C.M.**, Director, Resident Services and Family Programs, The Children's Inn at NIH
**Lucy Rector and Family**, Rare Disease Patient Family

10:20 a.m. **Session 1: Developing Treatments Through Innovative Trial Designs for Rare Diseases**
Innovative clinical trial designs are paving the way for more efficient and effective drug development, especially for complex and rare diseases. This session will introduce the topic of innovative trial designs. Presenters will discuss two case studies of currently ongoing clinical trials. This session also will discuss challenges for innovative trial designs.
**Moderator:** **Elena I. Schwartz, Ph.D.**, Program Officer, Coordinating Center for Clinical Trials, National Cancer Institute (NCI), NIH
**Panelists:**
- **Malcolm A. Smith, M.D., Ph.D.**, Associate Branch Chief for Pediatric Oncology, Clinical Investigations Branch, NCI, NIH
- **Troy Torgerson, M.D., Ph.D.**, Director of Experimental Immunology, Allen Institute for Immunology

10:50 a.m. **Break**
- Exhibits and Scientific Posters
- Art Exhibition
- Networking

11:10 a.m. **Rare Story: The Full-Time Job of Living Rare — Advocacy, Resilience and the Fight for Care**
**Lindsay Guentzel**, Multimedia Journalist, Storyteller, Advocate

11:30 a.m. **Session 2: Artificial Intelligence — Clinical Applications for Rare Diseases**
Artificial intelligence (AI) is revolutionizing how rare diseases challenges are addressed in research and regulation. This session introduces AI principles and tools and discusses their growing impact within the rare diseases ecosystem and limitations of use. Attendees will gain foundational knowledge and learn about AI use across research and clinical applications relevant to the rare diseases community. Presenters will then delve into two case studies that demonstrate AI use in rare diseases research.
**Moderator: Benjamin Soloman, M.D.**, Clinical Director, National Human Genome Research Institute, NIH
**Panelists:**
- **Nara Sobreira, M.D., Ph.D.**, Associate Professor in the McKusick-Nathans Department of Genetic Medicine and Associate Professor of Pediatrics, Johns Hopkins University
- **Vivek Rudrapatna, M.D., Ph.D.**, Assistant Professor in the Division of Gastroenterology and Director of the Center for Real-World Evidence, University of California, San Francisco
- **Tala Fakhouri, Ph.D., M.P.H.**, Vice President Consulting: AI & Digital Policy, Real-World Research, Parexel

12:30 p.m. **Lunch (on your own)**
- Exhibits and Scientific Posters
- Art Exhibition
- Networking

2:00 p.m. **Rare Research: How Collaboration Becomes Cures — A Juvenile Myositis and CAR T Story**
**Jim Minow**, Executive Director, Cure JM Foundation
**Leah Kania & Daughter Olivia**, Parent Advocate, Cure JM Foundation
**David Chang, M.D., M.P.H., FACR**, Chief Medical Officer, Cabaletta Bio

2:20 p.m. **Rare Story: Therapeutic Genome Editing for Hereditary Connective Tissue Disorders — A Patient-Scientist Perspective**
**Lauren Testa, Ph.D.**, Cell and Molecular Biology (Gene Therapy and Vaccines), University of Pennsylvania Perelman School of Medicine

2:40 p.m. **Rare Story: Conquering the Unconquerable — Turning Hope Into Progress Through Research**
**Jake Juip**, Rare Disease Advocate and College Student

3:00 p.m. **Rare Story: Jax's Story — The One-in-a-Million IPEX Fight for a Future**
**Melissa Ramirez, Ph.D.**, mom of Jax, IPEX Foundation Executive Director, Nationally Certified School Psychologist

3:20 p.m. **Break**
- Exhibits and Scientific Posters
- Art Exhibition
- Networking

3:40 p.m. **Session 3: Gene Therapy — Recent Approvals, Success Stories and Patient Perspectives**

Gene therapy continues to transform the treatment landscape for rare diseases, offering new hope to patients and families. This session will highlight two recent cases of gene therapy advancement and NCATS' involvement: the approval of a therapy for aromatic L-amino acid decarboxylase (AADC) deficiency and the development of a personalized CRISPR gene-editing therapy for severe carbamoyl phosphate synthetase 1 (CPS1) deficiency. The session also will include a broader discussion of patient considerations and decision-making factors when pursuing gene therapy.

**Moderator:** **Philip John (P.J.) Brooks, Ph.D.**, Acting Director, DRDRI, NCATS, NIH

**Panelists:**

- **Elizabeth A. Ottinger, Ph.D.**, Director, Therapeutic Development Branch, Division of Preclinical Innovation, NCATS, NIH
- **Rebecca Ahrens-Nicklas, M.D., Ph.D.**, Assistant Professor of Pediatrics, Division of Human Genetics, Children's Hospital of Philadelphia and the University of Pennsylvania
- **Kiran Musunuru, M.D., Ph.D., M.P.H., M.L., M.R.A.**, Professor of Cardiovascular Medicine, Genetics, and Pediatrics, Co-director, Orphan Disease Center, Perelman School of Medicine at the University of Pennsylvania
- **Nicole Muldoon**, Mother of Rare Disease Patient

4:40 p.m. **Closing Remarks**

**Joni L. Rutter, Ph.D.**, Director, NCATS, NIH

5:00 p.m. **Adjournment**

**Posters and Exhibits Close**